Table 2.1 Punnett square to predict genotype frequencies for loci on sex chromosomes and for all loci in males and females of haplo-diploid species. Notation in this table is based on birds where the sex chromosomes are Z and W (ZZ males and ZW females) with a diallelic locus on the Z chromosome possessing alleles A and a at frequencies p and q, respectively. In general, genotype frequencies in the homogametic or diploid sex are identical to Hardy–Weinberg expectations for autosomes, whereas genotype frequencies are equal to allele frequencies in the heterogametic or haploid sex.

Hemizygous or haploid sex			**Diploid sex**		
Genotype	Gamete	Frequency	Genotype	Gamete	Frequency
ZW	Z-A Z-a W	p q	ZZ	Z-A Z-a	p q
		Expected genotype frequencies under random mating			
Homogametic sex					
Z-A Z-A			p^2		
Z-A Z-a			$2pq$		
Z-a Z-a			q^2		
Heterogametic sex					
Z-A W			p		
Z-a W			q		

Table 2.2 Example DNA profile for three simple tandem repeat (STR) loci commonly used in human forensic cases. Locus names refer to the human chromosome (e.g. D3 means third chromosome) and chromosome region where the SRT locus is found.

Locus	D3S1358	D21S11	D18S51
Genotype	17, 18	29, 30	18, 18

Table 2.3 Allele frequencies for nine STR loci commonly used in forensic cases estimated from 196 US Caucasians sampled randomly with respect to geographic location. The allele names are the numbers of repeats at that locus (see Box 2.1). Allele frequencies (Freq) are as reported in Budowle et al. (2001), Table 1, from FBI sample population.

D3S1358		vWA		D21S11		D18S51		D13S317		FGA		D8S1179		D5S818		D7S820	
Allele	**Freq**	**Allele**	**Freq**	**Allele**	**Freq**	**Allele**	**Freq**	**Allele**	**Freq**	**Allele**	**Freq**	**Allele**	**Freq**	**Allele**	**Freq**	**Allele**	**Freq**
12	0.0000	13	0.0051	27	0.0459	<11	0.0128	8	0.0995	18	0.0306	<9	0.0179	9	0.0308	6	0.0025
13	0.0025	14	0.1020	28	0.1658	11	0.0128	9	0.0765	19	0.0561	9	0.1020	10	0.0487	7	0.0172
14	0.1404	15	0.1122	29	0.1811	12	0.1276	10	0.0510	20	0.1454	10	0.1020	11	0.4103	8	0.1626
15	0.2463	16	0.2015	30	0.2321	13	0.1224	11	0.3189	20.2	0.0026	11	0.0587	12	0.3538	9	0.1478
16	0.2315	17	0.2628	30.2	0.0383	14	0.1735	12	0.3087	21	0.1735	12	0.1454	13	0.1462	10	0.2906
17	0.2118	18	0.2219	31	0.0714	15	0.1276	13	0.1097	22	0.1888	13	0.3393	14	0.0077	11	0.2020
18	0.1626	19	0.0842	31.2	0.0995	16	0.1071	14	0.0357	22.2	0.0102	14	0.2015	15	0.0026	12	0.1404
19	0.0049	20	0.0102	32	0.0153	17	0.1556			23	0.1582	15	0.1097			13	0.0296
				32.2	0.1122	18	0.0918			24	0.1378	16	0.0128			14	0.0074
				33.2	0.0306	19	0.0357			25	0.0689	17	0.0026				
				35.2	0.0026	20	0.0255			26	0.0179						
						21	0.0051			27	0.0102						
						22	0.0026										

Table 2.4 Expected numbers of each of the three MN blood group genotypes under the null hypotheses of Hardy–Weinberg. Genotype frequencies are based on a sample of 1066 Chukchi individuals, a native people of eastern Siberia (Roychoudhury and Nei 1988).

Frequency of M = $\hat{p}$ = 0.4184
Frequency of N = $\hat{q}$ = 0.5816

Genotype	Observed	Expected number of genotypes	Observed – expected
MM	165	$N \times \hat{p}^2 = 1066 \times (0.4184)^2 = 186.61$	–21.6
MN	562	$N \times 2\hat{p}\hat{q} = 1066 \times 2(0.4184)(0.5816) = 518.80$	43.2
NN	339	$N \times \hat{q}^2 = 1066 \times (0.5816)^2 = 360.58$	–21.6

Table 2.5 χ^2 values and associated cumulative probabilities in the right-hand tail of the distribution for 1–5 df.

	Probability					
df	**0.5**	**0.25**	**0.1**	**0.05**	**0.01**	**0.001**
1	0.4549	1.3233	2.7055	3.8415	6.6349	10.8276
2	1.3863	2.7726	4.6052	5.9915	9.2103	13.8155
3	2.3660	4.1083	6.2514	7.8147	11.3449	16.2662
4	3.3567	5.3853	7.7794	9.4877	13.2767	18.4668
5	4.3515	6.6257	9.2364	11.0705	15.0863	20.5150

Table 2.6 Hardy–Weinberg expected genotype frequencies for the ABO blood groups under the hypotheses of (1) two loci with two alleles each and (2) one locus with three alleles. Both hypotheses have the potential to explain the observation of four blood group phenotypes. The notation fx is used to refer to the frequency of allele x. The underscore indicates any allele; for example, A_ means both AA and Aa genotypes. The observed blood type frequencies were determined for Japanese people living in Korea (from Berstein (1925) as reported in Crow (1993a)).

Blood type	Genotype		Expected genotype frequency		Observed (total = 502)
	Hypothesis 1	Hypothesis 2	Hypothesis 1	Hypothesis 2	
O	aa bb	OO	fa^2fb^2	fO^2	148
A	A_ bb	AA, AO	$(1 - fa^2)fb^2$	$fA^2 + 2fAfO$	212
B	aa B_	BB, BO	$fa^2(1 - fb^2)$	$fB^2 + 2fBfO$	103
AB	A_ B_	AB	$(1 - fa^2)(1 - fb^2)$	2fAfB	39

Table 2.7 Expected numbers of each of the four blood group genotypes under the hypotheses of (1) two loci with two alleles each and (2) one locus with three alleles. Estimated allele frequencies are based on a sample of 502 individuals.

Blood	Observed	Expected number of genotypes	Observed – expected	(Observed – expected)^2/ expected
Hypothesis 1 (fA = 0.293, fa = 0.707, fB = 0.153, fb = 0.847)				
O	148	$502(0.707)^2(0.847)^2 = 180.02$	–40.02	8.90
A	212	$502(0.500)(0.847)^2 = 180.07$	31.93	5.66
B	103	$502(0.707)^2(0.282) = 70.76$	32.24	14.69
AB	39	$502(0.500)(0.282) = 70.78$	–31.78	14.27
Hypothesis 2 (fA = 0.293, fB = 0.153, fO = 0.554)				
O	148	$502(0.554)^2 = 154.07$	–6.07	0.24
A	212	$502((0.293)^2 + 2(0.293)(0.554)) = 206.07$	5.93	0.17
B	103	$502((0.153)^2 + 2(0.153)(0.554)) = 96.85$	6.15	0.39
AB	39	$502(2(0.293)(0.153)) = 45.01$	–6.01	0.80

Table 2.8 Observed genotype counts and frequencies in a sample of $N = 200$ individuals for a single locus with two alleles. Allele frequencies in the population can be estimated from the genotype frequencies by summing the total count of each allele and dividing it by the total number of alleles in the sample ($2N$).

Genotype	Observed	Observed frequency	Allele count	Allele frequency
BB	142	$\frac{142}{200} = 0.71$	284 B	$\hat{p} = \frac{284 + 28}{400} = 0.78$
Bb	28	$\frac{28}{200} = 0.14$	28 B, 28 b	
bb	30	$\frac{30}{200} = 0.15$	60 b	$\hat{q} = \frac{60 + 28}{400} = 0.22$

Table 2.9 Estimates of the fixation index ($\hat{F}$) for various species and breeds based on pedigree or molecular genetic marker data.

Species	Mating system	$\hat{F}$	Method	Reference
Human				
Homo sapiens	Outcrossed	0.0001–0.046	Pedigree	Jorde 1997
Snail				
Bulinus truncates	Selfed and outcrossed	0.6–1.0	Microsatellites	Viard et al. 1997
Domestic dogs				
Breeds combined	Outcrossed	0.33	Allozyme	Christensen et al. 1985
German Shepherd	Outcrossed	0.10		
Mongrels	Outcrossed	0.06		
Plants				
Arabidopsis thaliana	Selfed	0.99	Allozyme	Abbott and Gomes 1989
Pinus ponderosa	Outcrossed	–0.37	Allozyme	Brown 1979

Table 2.10 The mean phenotype in a population that is experiencing consanguineous mating. The inbreeding coefficient is f and $d = 0$ when there is no dominance.

Genotype	Phenotype	Frequency	Contribution to population mean
AA	$+a$	$p^2 + fpq$	$ap^2 + afpq$
Aa	d	$2pq - f2pq$	$d2pq - df2pq$
aa	$-a$	$q^2 + fpq$	$-aq^2 - afpq$

Population mean: $ap^2 + d2pq - df2pq - aq^2 = a(p - q) + d2pq(1 - f)$

Table 2.11 A summary of the Mendelian basis of inbreeding depression under the dominance and overdominance hypotheses along with predicted patterns of inbreeding depression with continued consanguineous mating.

Hypothesis	Mendelian basis	Low-fitness genotypes	Changes in inbreeding depression with continued consanguineous mating
Dominance	Recessive and partly recessive deleterious alleles	Only homozygotes for deleterious recessive alleles	Purging of deleterious alleles that is increasingly effective as degree of recessiveness increases
Overdominance	Heterozygote advantage or heterosis	All homozygotes	No changes as long as consanguineous mating keeps heterozygosity low

Table 2.12 Expected frequencies of gametes for two diallelic loci in a randomly mating population with a recombination rate between the loci of *r*. The first eight genotypes have non-recombinant and recombinant gametes that are identical. The last two genotypes produce novel recombinant gametes, requiring inclusion of the recombination rate to predict gamete frequencies. Summing down each column of the table gives the total frequency of each gamete in the next generation.

		Frequency of gametes in next generation			
Genotype	**Expected frequency**	**A_1B_1**	**A_2B_2**	**A_1B_2**	**A_2B_1**
A_1B_1/A_1B_1	$(p_1q_1)^2$	$(p_1q_1)^2$			
A_2B_2/A_2B_2	$(p_2q_2)^2$		$(p_2q_2)^2$		
A_1B_1/A_1B_2	$2(p_1q_1)(p_1q_2)$	$(p_1q_1)(p_1q_2)$		$(p_1q_1)(p_1q_2)$	
A_1B_1/A_2B_1	$2(p_1q_1)(p_2q_1)$	$(p_1q_1)(p_2q_1)$			$(p_1q_1)(p_2q_1)$
A_2B_2/A_1B_2	$2(p_2q_2)(p_1q_2)$		$(p_2q_2)(p_1q_2)$	$(p_2q_2)(p_1q_2)$	
A_2B_2/A_2B_1	$2(p_2q_2)(p_2q_1)$		$(p_2q_2)(p_2q_1)$		$(p_2q_2)(p_2q_1)$
A_1B_2/A_1B_2	$(p_1q_2)^2$			$(p_1q_2)^2$	
A_2B_1/A_2B_1	$(p_2q_1)^2$				$(p_2q_1)^2$
A_2B_2/A_1B_1	$2(p_2q_2)(p_1q_1)$	$(1-r)(p_2q_2)(p_1q_1)$	$(1-r)(p_2q_2)(p_1q_1)$	$r(p_2q_2)(p_1q_1)$	$r(p_2q_2)(p_1q_1)$
A_1B_2/A_2B_1	$2(p_1q_2)(p_2q_1)$	$r(p_1q_2)(p_2q_1)$	$r(p_1q_2)(p_2q_1)$	$(1-r)(p_1q_2)(p_2q_1)$	$(1-r)(p_1q_2)(p_2q_1)$

Table 2.13 Example of the effect of population admixture on gametic disequilibrium. In this case the two populations are each at gametic equilibrium given their respective allele frequencies. When an equal number of gametes from each of these two genetically diverged populations are combined to form a new population, gametic disequilibrium results from the diverged gamete frequencies in the founding populations. The allele frequencies are: population 1 $p_1 = 0.1$, $p_2 = 0.9$, $q_1 = 0.1$, $q_2 = 0.9$; population 2 $p_1 = 0.9$, $p_2 = 0.1$, $q_1 = 0.9$, $q_2 = 0.1$. In population 1 and population 2 gamete frequencies are the product of their respective allele frequencies as expected under independent segregation. In the mixture population, all allele frequencies become the average of the two source populations (0.5) with $D_{max} = 0.25$.

Gamete/*D*	Gamete frequency	Population 1	Population 2	Mixture population
A_1B_1	g_{11}	0.01	0.81	0.41
A_2B_2	g_{22}	0.81	0.01	0.41
A_1B_2	g_{12}	0.09	0.09	0.09
A_2B_1	g_{21}	0.09	0.09	0.09
D		0.0	0.0	0.16
D′		0.0	0.0	0.16/0.25 = 0.64

Table 2.14 Joint counts of genotype frequencies observed at two microsatellite loci in the fish *Morone saxatilis*. Alleles at each locus are indicated by numbers (e.g. 12 is a heterozygote and 22 is a homozygote).

	Genotype at locus AC25-6#10					
Genotype at locus AT150-2#4	**12**	**22**	**33**	**24**	**44**	**Row totals**
22	0	0	1	0	0	1
24	1	4	0	4	1	10
44	2	15	0	0	0	17
25	0	3	0	0	0	3
45	0	8	0	1	0	9
55	1	1	0	0	0	2
26	0	1	0	2	0	3
46	1	3	0	0	0	4
56	0	0	0	1	0	1
Column totals	5	35	1	8	1	50

	$N = 4$			$N = 20$		
Trial	**Blue**	**Clear**	***p***	**Blue**	**Clear**	***p***
1	1	3	0.25	12	8	0.60
2	2	2	0.50	10	10	0.50
3	3	1	0.75	9	11	0.45
4	0	4	0.0	7	13	0.35
5	2	2	0.50	8	12	0.40
6	1	3	0.25	11	9	0.55
7	2	2	0.50	11	9	0.55
8	3	1	0.75	12	8	0.60
9	2	2	0.50	10	10	0.50
10	1	3	0.25	9	11	0.45

Table 3.1 Typical results of sampling from beaker populations of micro-centrifuge tubes where the frequency of both blue (*p*) and clear tubes is $^1/_2$. After each draw, all tubes are replaced and the beaker is mixed to randomize the tubes for the next draw.

Table 3.2 The equations used to calculate the expected frequency of populations with zero, one, or two A alleles in generation one ($t = 1$) based on the previous generation ($t = 0$). Frequencies at $t = 1$ depend both on transition probabilities due to sampling error (constant terms like 0, 1, or $^1/_2$) and population frequencies in the previous generation ($P_{t=0}(x)$ terms). The transition probabilities are calculated with the binomial formula $\left(P_{i\to j} = \left(\frac{2N}{j}\right)p^j q^{2N-j}\right)$. Since sampling error cannot change the allele frequency of a population at fixation or loss, $P_{2\to2} = 1$ and $P_{0\to0} = 1$, whereas the other possibilities have a probability of zero.

One generation later ($t = 1$)			**Initial state: number of A alleles ($t = 0$)**				
A alleles	**Expected frequency**		**2**		**1**		**0**
2	$P_{t=1}(2)$	=	$(P_{2\to2})P_{t=0}(2)$	+	$(P_{1\to2})P_{t=0}(1)$	+	$(0)P_{t=0}(0)$
1	$P_{t=1}(1)$	=	$(0)P_{t=0}(2)$	+	$(P_{1\to1})P_{t=0}(1)$	+	$(0)P_{t=0}(0)$
0	$P_{t=1}(0)$	=	$(0)P_{t=0}(2)$	+	$(P_{1\to0})P_{t=0}(1)$	+	$(P_{0\to0})P_{t=0}(0)$

Table 3.3 Levels of heterozygosity found in island and mainland populations of the same species demonstrates that small population size has effects akin to inbreeding. Heterozygosity in island and mainland populations is compared using the effective inbreeding coefficient $F_e = 1 - \frac{H_{island}}{H_{mainland}}$. $F_e > 0$ when the mainland population(s) exhibit more heterozygosity, $F_e < 0$ when the island population(s) exhibit more heterozygosity, and F_e is 0 when levels of heterozygosity are equal. Values given are ranges when more than one set of comparisons was reported from a single source. Data from Frankham (1998).

Species	F_e
Mammals	
Wolf (*Canis lupis*)	0.552
Lemur (*Lemur macaco*)	0.518
Mouse (*Mus musculus*)	–0.048 to 1.000
Norway rat (*Rattus rattus*)	–0.355 to 0.710
Leopard (*Panthera pardus*)	0.548
Cactus mouse (*Peromyscus eremicus*)	0.445–0.899
Shrew (*Sorex cinereus*)	–0.241 to 0.468
Black bear (*Ursus americanus*)	0.545
Birds	
Singing starling (*Aplonis cantoroides*)	0.231–0.833
Chaffinch (*Fringilla coelebs*)	–0.164 to 0.504
Reptiles	
Shingleback lizard (*Trachydosaurus rugosus*)	0.069–0.311

Table 3.4 Data from simulated allele frequencies in Fig. 3.20 used to estimate the effective population size. Here, the ratio of heterozygosity in generations three and four is used to estimate inbreeding effective population size ($\hat{N}_e^i$) according to equation 3.59. Initial allele frequencies were $p = q = 0.5$, so $H_{t=1} = 0.5$. One generation of genetic drift took place, hence 1 is used in the numerator of the expression for N_e^i. The average N_e^i excludes the negative values.

$H_{t=3}$	$H_{t=4}$	$\ln\left(\frac{H_{t=4}}{H_{t=3}}\right)$	$\hat{N}_e^i = -\frac{1}{2}\frac{1}{\ln\left(\frac{H_{t=4}}{H_{t=3}}\right)}$
0.4987	0.4504	–0.1018	4.91
0.4866	0.4594	–0.0575	8.69
0.4813	0.3474	–0.3259	1.53
0.4998	0.4376	–0.1329	3.76
0.4546	0.3772	–0.1864	2.68
0.4884	0.4999	0.0232	–21.58
0.4920	0.4566	–0.0747	6.69
0.4413	0.4856	0.0957	–5.22
0.4715	0.3578	–0.2761	1.81
0.4995	0.4550	–0.0932	5.36
			Average $\hat{N}_e^i = 4.43$

Table 3.5 Data from simulated allele frequencies in Fig. 3.20 used to estimate the effective population size. Here, the change in allele frequency between generations three and four is used to estimate variance effective population size ($\hat{N}_e^v$) according to equation 3.56. Allele frequencies in the third generation were used to estimate pq.

$p_{t=4}$	$\Delta p = p_{t=4} - p_{t=3}$	pq	$\text{Var}(\Delta p) = \frac{1}{10}\sum(p_{t=4} - \bar{p})^2$	$\hat{N}_e^v = \frac{pq}{2 \times \text{variance}(\Delta p)}$
0.6574	0.1825	0.2494	0.0186	6.71
0.3575	–0.0606	0.2433		6.55
0.2238	–0.1795	0.2406		6.47
0.3234	–0.1668	0.2499		6.72
0.2523	–0.0970	0.2273		6.12
0.4940	–0.0819	0.2442		6.57
0.6473	0.0842	0.2460		6.62
0.4153	0.0866	0.2207		5.94
0.7667	0.1473	0.2357		6.34
0.6499	0.1343	0.2498		6.72
				Average $\hat{N}_e^v = 6.48$

Table 3.6 Estimates of the ratio of effective to census population size $\left(\frac{N_e}{N}\right)$ for various species based on a wide range of estimation methods and assumptions.

Species	$\frac{N_e}{N}$	Reference
Leopard frog (*Rana pipiens*)	0.1–1.0	Hoffman et al. 2004
New Zealand snapper (*Pagrus auratus*)	$(0.25–16.7) \times 10^{-5}$	Hauser et al. 2002
Red drum (*Sciaenops ocellatus*)	0.001	Turner et al. 2002
White-toothed shrew (*Crocidura russula*)	0.60	Bouteiller and Perrin 2000
Flour beetle (*Tribolium castaneum*)	0.81–1.02[a]	Pray et al. 1996
Review of 102 species	0.10[b]	Frankham 1995

[a]Ratios declined as census population sizes increased.
[b]Mean of 56 estimates in the "comprehensive data set" that included impacts of unequal breeding sex ratio, variance in family size, and fluctuating population size over time.

Table 4.1 Microsatellite genotypes (given in base pairs) for some of the 30 mature individuals of the tropical tree *Corythophora alta* sampled from a 9 ha plot of continuous forest in the Brazilian Amazon. Progeny are seeds collected from known trees. Missing data are indicated by a 0.

	Genotype									
Microsatellite locus . . .	**A**		**B**		**C**		**D**		**E**	
Candidate parents										
684	333	339	97	106	169	177	275	305	135	135
989	330	336	97	106	165	181	275	275	135	153
1072	315	333	103	106	169	179	296	302	138	138
1588	318	327	106	106	165	167	272	293	135	150
1667	324	333	0	0	165	185	275	284	141	159
1704	318	327	103	106	0	0	284	296	144	147
1836	333	339	97	97	181	183	275	296	138	144
1946	327	333	91	106	167	187	284	287	135	147
2001	321	336	0	0	177	181	284	302	138	144
2121	318	333	100	106	179	181	284	302	144	144
2395	327	333	103	103	179	187	275	296	150	159
3001	324	333	91	106	167	183	284	302	147	159
3226	327	327	103	106	163	181	275	275	135	144
3237	324	324	91	103	179	187	284	305	144	159
3547	321	321	103	106	177	179	275	296	0	0
4112	327	327	97	106	169	181	296	302	144	144
4783	321	327	0	0	183	185	290	308	144	156
4813	327	333	106	106	177	179	284	302	135	138
4865	321	327	106	106	167	179	284	296	144	153
4896	315	333	100	106	181	189	275	284	162	162
5024	318	327	100	103	165	167	275	284	147	147
Seed progeny										
989 seed 1-1	327	336	91	106	165	185	275	287	153	153
989 seed 2-1	327	330	103	106	165	181	275	275	135	135
989 seed 3-1	330	336	97	106	165	181	0	0	135	153
989 seed 25-1	321	330	106	106	167	181	275	296	135	153

Table 4.2 Seed progeny genotypes (top row of every three) given with the known maternal parent genotype (middle row of every three) along with the genotype of the most probable paternal parent (bottom row of every three) from the pool of all possible candidate parents. Alleles in the seed progeny that match those in the known maternal parent are underlined. The known maternal parent is also a candidate paternal parent since this species can self-fertilize. Missing data are indicated by zero.

	Genotype									
Microsatellite locus . . .	**A**		**B**		**C**		**D**		**E**	
989 seed 1-1	327	336	91	106	165	185	275	287	153	153
989	330	336	97	106	165	181	275	275	135	153
1946	327	333	91	106	167	185	284	287	135	147
989 seed 2-1	327	330	103	106	165	181	275	275	135	135
989	330	336	97	106	165	181	275	275	135	153
3226	327	327	103	106	163	181	275	275	135	144
989 seed 3-1	330	336	97	106	165	181	0	0	135	153
989	330	336	97	106	165	181	275	275	135	153
989	330	336	97	106	165	181	275	275	135	153
989 seed 25-1	321	330	106	106	167	181	275	296	135	153
989	330	336	97	106	165	181	275	275	135	153
4865	321	327	106	106	167	179	284	296	144	153

Table 4.3 Allele frequencies for five *Corythophora alta* microsatellite loci used for paternity analysis.

Microsatellite locus . . .	A		B		C		D		E	
	Allele	Frequency	Allele	Frequency	Allele	Frequency	Allele	Frequency	Allele	Frequency
	315	0.0405	91	0.0735	163	0.0217	272	0.0238	135	0.2917
	318	0.0541	97	0.3088	165	0.2283	275	0.4167	138	0.0625
	321	0.1216	100	0.0735	167	0.0761	281	0.0357	141	0.0313
	324	0.0541	103	0.1471	169	0.0435	284	0.1429	144	0.2188
	327	0.2703	106	0.3971	171	0.0217	287	0.0119	147	0.0625
	330	0.1892			177	0.0543	290	0.0119	150	0.0938
	333	0.1216			179	0.1304	293	0.0238	153	0.1250
	336	0.1216			181	0.2065	296	0.1905	156	0.0208
	339	0.0270			183	0.0652	299	0.0119	159	0.0521
					185	0.0435	302	0.0833	162	0.0417
					187	0.0326	305	0.0357		
					189	0.0109	308	0.0119		
					193	0.0109				
					197	0.0543				

Table 4.4 The chance of a random match for the included fathers in Table 4.2. The probability of a random match at each locus is $p_i^2 + 2p_i(1 - p_i)$. The combined probability of a random match for all loci in the haplotype is the product of the probabilities of a random match at each independent locus. Paternal haplotype data are treated as missing (0) for the purposes of probability calculations when progeny genotype data are missing. In the cases where the paternal haplotype has multiple possible alleles at some loci, the highest probability of a chance match is given. The allele frequencies for each locus are given in Table 4.3.

	Microsatellite haplotype									
Included father	**A**		**B**		**C**		**D**	**E**		***P*(multilocus random match)**
1946 (seed 1-1)	327		91		185		287	135		
Allele frequencies	0.2703		0.0735		0.0435		0.0119	0.2917		
P(random match)	0.4675		0.1416		0.0851		0.0237	0.4983		0.0000665
3226 (seed 2-1)	327		103	106	181	275	135			
Allele frequencies	0.2703		0.0735	0.3971	0.2065	0.4167	0.2917			
P(random match)	0.4675		0.1416	0.6365	0.3704	0.6598	0.4983			≤0.03624
989 (seed 3-1)	330	336	97	106	165	181	0	135	153	
Allele frequencies	0.1892	0.1216	0.3088	0.3971	0.2283	0.2065	1.0	0.2917	0.1250	
P(random match)	0.3426	0.2284	0.5222	0.6365	0.4045	0.3704	1.0	0.4983	0.2344	≤0.0440

Table 4.5 The mathematical and biological definitions of heterozygosity for three levels of population organization. In the summations, *i* refers to each subpopulation 1, 2, 3 . . . *n* and p_i and q_i are the frequencies of the two alleles at a diallelic locus in subpopulation *i*.

$H_I = \frac{1}{n}\sum_{i=1}^{n} \hat{H}_i$	The average observed heterozygosity within each subpopulation.
$H_S = \frac{1}{n}\sum_{i=1}^{n} 2p_i q_i$	The average expected heterozygosity of subpopulations assuming random mating within each subpopulation.
$H_T = 2\bar{p}\bar{q}$	The expected heterozygosity of the total population assuming random mating within subpopulations and no divergence of allele frequencies among subpopulations.

Table 4.6 The mathematical and biological definitions of fixation indices for two levels of population organization.

$F_{IS} = \frac{H_S - \bar{H}_I}{H_S}$	The average difference between observed and Hardy–Weinberg expected heterozygosity within each subpopulation due to non-random mating. The correlation between the states of two alleles in a genotype sampled at random from any subpopulation.
$F_{ST} = \frac{H_T - H_S}{H_T}$	The reduction in heterozygosity due to subpopulation divergence in allele frequency. The difference between the average expected heterozygosity of subpopulations and the expected heterozygosity of the total population. Alternately, the probability that two alleles sampled at random from a single subpopulation are identical given the probability that two alleles sampled from the total population are identical.
$F_{IT} = \frac{H_T - H_I}{H_T}$	The correlation between the states of two alleles in a genotype sampled at random from a single subpopulation given the possibility of non-random mating within populations *and* allele frequency divergence among populations.

Table 4.7 Allele and genotype frequencies for the hypothetical example of albino squirrels in Fig. 4.11 used to demonstrate Wahlund's principle. Initially, the total population is subdivided into two demes with different allele frequencies. These two populations are then fused and undergo one generation of random mating.

	Initial subpopulations	Fused population
Allele frequency q	0.4 and 0.0	$\frac{0.4+0.0}{2} = 0.2$
Variance in q	$\frac{(0.4-0.2)^2+(0.0-0.2)^2}{2} = 0.04$	0
Frequency of aa	$\overline{q^2} = \frac{0.16+0}{2} = 0.08$	$(0.2)^2 = 0.04$
Frequency of Aa	$\overline{2pq} = \frac{0.48+0.0}{2} = 0.24$	$2(0.2)(0.8) = 0.32$
Frequency of AA	$\overline{p^2} = \frac{0.36+1.0}{2} = 0.68$	$(0.8)^2 = 0.64$

Table 4.8 Expected frequencies for individual DNA-profile loci and the three loci combined with and without adjustment for population structure. Calculations assume that $F_{IS} = 0$ and use the upper-bound estimate of $F_{ST} = 0.05$ in human populations. Allele frequencies are given in Table 2.3.

	Expected genotype frequency	
Locus	**With panmixia**	**With population structure**
D3S1358	$2(0.2118)(0.1626) = 0.0689$	$2(0.2118)(0.1626)(1 - 0.05) = 0.0655$
D21S11	$2(0.1811)(0.2321) = 0.0841$	$2(0.1811)(0.2321)(1 - 0.05) = 0.0799$
D18S51	$(0.0918)^2 = 0.0084$	$(0.0918)^2 + 0.0918(1 - 0.0918)(0.05) = 0.0126$
All loci	$(0.0689)(0.0841)(0.0084) = 0.000049$	$(0.0655)(0.0799)(0.0126) = 0.000066$

Table 4.9 Estimates of the fixation index among subpopulations ($\hat{F}_{ST}$) for diverse species based on molecular genetic marker data for nuclear loci. Different estimators were employed depending on the type of genetic marker and study design. Each $\hat{F}_{ST}$ was used to determine the effective number of migrants ($\widehat{N_e m}$) that would produce an identical level of population structure under the assumptions of the infinite island model according to equation 4.63.

Species	$\hat{F}_{ST}$	$\widehat{N_e m}$	Reference
Amphibians			
Alytes muletansis (Mallorcan midwife toad)	0.12–0.53	1.8–0.2	Kraaijeveld-Smit et al. 2005
Birds			
Gallus gallus (broiler chicken breed)	0.19	1.0	Emara et al. 2002
Mammals			
Capreolus capreolus (roe deer)	0.097–0.146	2.2–1.4	Wang and Schreiber 2001
Homo sapiens (human)	0.03–0.05	7.8–4.6	Rosenberg et al. 2002
Microtus arvalis (common vole)	0.17	1.2	Heckel et al. 2005
Plants			
Arabidopsis thaliana (mouse-ear cress)	0.643	0.1	Bergelson et al. 1998
Oryza officinalis (wild rice)	0.44	0.3	Gao 2005
Phlox drummondii (annual phlox)	0.17	1.2	Levin 1977
Prunus armeniaca (apricot)	0.32	0.5	Romero et al. 2003
Fish			
Morone saxatilis (striped bass)	0.002	11.8	Brown et al. 2005
Sparisoma viride (stoplight parrotfish)	0.019	12.4	Geertjes et al. 2004
Insects			
Drosophila melanogaster (fruit fly)	0.112	2.0	Singh and Rhomberg 1987
Glossina pallidipes (tsetse fly)	0.18	1.1	Ouma et al. 2005
Heliconius charithonia (butterfly)	0.003	79.8	Kronforst and Flemming 2001
Corals			
Seriatopora hystrix	0.089–0.136	2.6–1.6	Maier et al. 200

Table 5.1 Per-locus mutation rates measured for five loci that influence coat-color phenotypes in inbred lines of mice (Schlager & Dickie 1971). Dominant mutations were counted by examining the coat color of F1 progeny from brother–sister matings. Recessive mutations required examining the coat color of F1 progeny from crosses between an inbred line homozygous for a recessive allele and a homozygous wild-type dominant allele. The effort to obtain these estimates was truly incredible, involving around 7 million mice observed over the course of 6 years.

Locus	Gametes tested	Mutations observed	Mutation rate per locus × 10^{-6} (95% CI)
Mutations from dominant to recessive alleles			
Albino	150,391	5	33.2 (10.8–77.6)
Brown	919,699	3	3.3 (0.7–9.5)
Dilute	839,447	10	11.9 (5.2–21.9)
Leaden	243,444	4	16.4 (4.5–42.1)
Non-agouti	67,395	3	44.5 (9.2–130.1)
All loci	2,220,376	25	11.2 (7.3–16.6)
Mutations from recessive to dominant alleles			
Albino	3,423,724	0	0 (0.0–1.1)
Brown	3,092,806	0	0 (0.0–1.2)
Dilute	2,307,692	9	3.9 (1.8–11.1)
Leaden	266,122	0	0 (0.0–13.9)
Non-agouti	8,167,854	34	4.2 (2.9–5.8)
All loci	17,236,978	43	2.5 (1.8–3.4)

95% CI, 95% confidence interval.

Table 5.2 Rates of spontaneous mutation expressed per genome and per base pair for a range of organisms. The most reliable estimates come from microbes with DNA genomes whereas estimates from RNA viruses and eukaryotes have greater uncertainty. Full explanation of the assumptions and uncertainties behind these estimates can be found in Drake et al. (1998).

Organism	Mutation rate per replication	
	Per genome	Per base pair
Lytic RNA viruses		
Bacteriophage Qβ	6.5	
Poliovirus	0.8	
Vesicular stomatitis virus	3.5	
Influenza A	≥1.0	
Retroviruses		
Spleen necrosis virus	0.04	
Rous sarcoma virus	0.43	
Bovine leukemia virus	0.027	
Human immunodeficiency virus	0.16–0.22	
DNA-based microbes		
Bacteriophage M13	0.0046	7.2×10^{-7}
Bacteriophage λ	0.0038	7.7×10^{-8}
Bacteriophages T2 and T4	0.0040	2.4×10^{-8}
Escherichia coli	0.0025	5.4×10^{-10}
Neurospora crassa	0.0030	7.2×10^{-11}
Saccharomyces cerevisiae	0.0027	2.2×10^{-10}
Eukaryotes		
Caenorhabditis elegans	0.018	2.3×10^{-10}
Drosophila	0.058	3.4×10^{-10}
Human	0.49	1.8×10^{-10}
		2.5×10^{-8}[a]
Mouse	0.16	5.0×10^{-11}

[a]Estimate from Nachman and Crowell (2000) based on pseudogene divergence between humans and chimpanzees.

Table 5.3 The expected frequency of each family size per pair of parents (k) under the Poisson distribution with a mean family size of 2 ($\bar{k} = 2$). Also given is the expected probability that a mutant allele A_m would not be transmitted to any progeny for a given family size. Note that 0! equals one.

Family size per pair of parents (k) . . .	**0**	**1**	**2**	**3**	**4 . . . k**
Expected frequency	e^{-2}	$2e^{-2}$	$2e^{-2}$	$\frac{4}{3}e^{-2}$	$\frac{2}{3}e^{-2} \ldots \frac{2^k}{k!}e^{-2}$
Chance that A_m is not transmitted	1	$\frac{1}{2}$	$\left(\frac{1}{2}\right)^2$	$\left(\frac{1}{2}\right)^3$	$\left(\frac{1}{2}\right)^4 \ldots \left(\frac{1}{2}\right)^k$

Table 5.4 Hypothetical allele frequencies in two subpopulations used to compute the standard genetic distance, *D*. This example assumes three alleles at one locus, but loci with any number of alleles can be used. *D* for multiple loci uses the averages of J_{11}, J_{22}, and J_{12} for all loci to compute the genetic identity *I*.

Allele	Subpopulation 1		Subpopulation 2	
	Frequency	p_{ik}^2	Frequency	p_{ik}^2
1	0.60	$p_{11}^2 = 0.36$	0.40	$p_{21}^2 = 0.16$
2	0.30	$p_{12}^2 = 0.09$	0.60	$p_{22}^2 = 0.36$
3	0.10	$p_{13}^2 = 0.01$	0.00	$p_{23}^2 = 0.00$

Table 5.5 A comparison of hypothetical estimates of population subdivision assuming the infinite alleles model using F_{ST} or assuming the stepwise mutation model using R_{ST}. Allelic data expressed as the number of repeats at a hypothetical microsatellite locus are given for two subpopulations in each of two cases. In the case on the left, the majority of alleles in both populations are very similar in state. Under the stepwise mutation model the two alleles are separated by a single change that could be due to mutation. The estimate of R_{ST} is therefore less than the estimate of F_{ST}. In the case on the right, the two populations have alleles that are very different in state and more than a single mutational change apart under the stepwise mutation model. In contrast, all alleles are a single mutational event apart in the infinite alleles model. The higher estimate of R_{ST} reflects greater weight given to larger differences in allelic state.

	Case 1	Case 2
Subpopulation 1 (number of repeats)	9, 10, 10, 10, 10, 10, 10, 10, 10, 10	9, 10, 10, 10, 10, 10, 10, 10, 10, 10
Subpopulation 2 (number of repeats)	12, 11, 11, 11, 11, 11, 11, 11, 11, 11	19, 20, 20, 20, 20, 20, 20, 20, 20, 20
Allele size variance in subpopulation 1, S_1	0.10	0.10
Allele size variance in subpopulation 2, S_2	0.10	0.10
Allele size variance in total population, S_T	0.947	52.821
R_{ST}	0.789	0.996
Expected heterozygosity in subpopulation 1, H_1	0.18	0.18
Expected heterozygosity in subpopulation 2, H_2	0.18	0.18
Average subpopulation expected heterozygosity, H_S	0.18	0.18
Expected heterozygosity in total population, H_T	0.59	0.59
F_{ST}	0.695	0.695

Table 6.1 The expected frequencies of two genotypes after natural selection, for the case of clonal reproduction. The top section of the table gives expressions for the general case. The bottom part of the table uses absolute and relative fitness values identical to Fig. 6.1 to show the change in genotype proportions for the first generation of natural selection. The absolute fitness of the A genotype is highest and is therefore used as the standard of comparison when determining relative fitness.

	Genotype	
	A	**B**
Generation *t*		
Initial frequency	p_t	q_t
Genotype-specific growth rate (absolute fitness)	λ_A	λ_B
Relative fitness	$w_A = \frac{\lambda_A}{\lambda_A}$	$w_B = \frac{\lambda_B}{\lambda_A}$
Frequency after natural selection	$p_t w_A$	$q_t w_B$
Generation *t* + 1		
Initial frequency p_{t+1}	$\frac{p_t w_A}{p_t w_A + q_t w_B}$	$\frac{q_t w_B}{p_t w_A + q_t w_B}$
Change in genotype frequency	$\Delta p = p_{t+1} - p_t$	$\Delta q = q_{t+1} - q_t$
Generation *t*		
Initial frequency	$p_t = 0.5$	$q_t = 0.5$
Genotype-specific growth rate (absolute fitness)	$\lambda_A = 1.03$	$\lambda_B = 1.01$
Relative fitness	$w_A = \frac{\lambda_A}{\lambda_A} = \frac{1.03}{1.03} = 1.0$	$w_B = \frac{\lambda_B}{\lambda_A} = \frac{1.01}{1.03} = 0.981$
Frequency after natural selection	$p_t w_A = (0.5)(1.0) = 0.5$	$q_t w_B = (0.5)(0.981) = 0.4905$
Generation *t* + 1		
Initial frequency p_{t+1}	$\frac{0.5}{0.5 + 0.4905} = 0.5048$	$\frac{0.4905}{0.5 + 0.4905} = 0.4952$
Change in genotype frequency	$0.5048 - 0.5 = 0.0048$	$0.4952 - 0.05 = -0.0048$

Table 6.2 Assumptions of the basic natural selection model with a diallelic locus.

Genetic
- Diploid individuals
- One locus with two alleles
- Obligate sexual reproduction

Reproduction
- Generations do not overlap
- Mating is random

Natural selection
- Mechanism of natural selection is genotype-specific differences in survivorship (fitness) that lead to variable genotype-specific growth rates, termed viability selection
- Fitness values are constants that do not vary with time, over space, or in the two sexes

Population
- Infinite population size so there is no genetic drift
- No population structure
- No gene flow
- No mutation

Table 6.3 The expected frequencies of three genotypes after natural selection for a diallelic locus with sexual reproduction and random mating. The absolute fitness of the AA genotype is used as the standard of comparison when determining relative fitness.

	Genotype		
	AA	**Aa**	**aa**
Generation t			
Initial frequency	p_t^2	$2p_tq_t$	q_t^2
Genotype-specific survival (absolute fitness)	ℓ_{AA}	ℓ_{Aa}	ℓ_{aa}
Relative fitness	$w_{AA} = \frac{\ell_{AA}}{\ell_{AA}}$	$w_{Aa} = \frac{\ell_{Aa}}{\ell_{AA}}$	$w_{aa} = \frac{\ell_{aa}}{\ell_{AA}}$
Frequency after natural selection	$p_t^2 w_{AA}$	$2p_tq_tw_{Aa}$	$q_t^2 w_{aa}$
Average fitness	$p_t^2 w_{AA} + 2p_tq_tw_{Aa} + p_t^2 w_{aa}$		
Generation $t+1$			
Genotype frequency	$\frac{p_t^2 w_{AA}}{\bar{w}}$	$\frac{2p_tq_tw_{Aa}}{\bar{w}}$	$\frac{q_t^2 w_{aa}}{\bar{w}}$
Allele frequency	$p_{t+1} = \frac{p_t(p_tw_{AA} + q_tw_{Aa})}{\bar{w}}$	$q_{t+1} = \frac{q_t(q_tw_{aa} + p_tw_{Aa})}{\bar{w}}$	
Change in allele frequency	$\Delta p = \frac{pq[p(w_{AA} - w_{Aa}) + q(w_{Aa} - w_{aa})]}{\bar{w}}$	$\Delta q = \frac{pq[q(w_{aa} - w_{Aa}) + p(w_{Aa} - w_{AA})]}{\bar{w}}$	

Table 6.4 The general categories of relative fitness values for viability selection at a diallelic locus. The variables *s* and *t* are used to represent the decrease in viability of a genotype compared to the maximum fitness of 1 ($1 - w_{xx} = s$). The degree of dominance of the A allele is represented by *h* with additive gene action (sometime called codominance) when $h = {}^1/_2$.

Category	Genotype-specific fitness		
	w_{AA}	w_{Aa}	w_{aa}
Selection against a recessive phenotype	1	1	$1 - s$
Selection against a dominant phenotype	$1 - s$	$1 - s$	1
General dominance (dominance coefficient $0 \leq h \leq 1$)	1	$1 - hs$	$1 - s$
Heterozygote disadvantage (underdominance for fitness)	1	$1 - s$	1
Heterozygote advantage (overdominance for fitness)	$1 - s$	1	$1 - t$

Table 7.1 Relative fitness estimates for the six genotypes of the hemoglobin β gene estimated in Western Africa where malaria is common. Values from Cavallo-Sforza and Bodmer (1971) are based by deviation from Hardy–Weinberg expected genotype frequencies. Values from Hedrick (2004) are estimated from relative risk of mortality for individuals with AA, AC, AS, and CC genotypes and assume 20% overall mortality from malaria.

	Relative fitness (w)					
Genotype . . .	**AA**	**AS**	**SS**	**AC**	**SC**	**CC**
From Cavallo-Sforza and Bodmer (1971)						
Relative to w_{CC}	0.679	0.763	0.153	0.679	0.534	1.0
Relative to w_{AS}	0.89	1.0	0.20	0.89	0.70	1.31
From Hedrick (2004)						
Relative to w_{CC}	0.730	0.954	0.109	0.865	0.498	1.0
Relative to w_{AS}	0.765	1.0	0.114	0.906	0.522	1.048

Table 7.2 Matrix of fitness values for all combinations of the four gametes formed at two diallelic loci (top). If the same gamete inherited from either parent has the same fitness in a progeny genotype (e.g. $w_{12} = w_{21}$), then there are 10 gamete fitness values shown outside the shaded triangle. These 10 fitness values can be summarized by a genotype fitness matrix (bottom) under the assumption that double heterozygotes have equal fitness ($w_{14} = w_{23}$) and representing their fitness value by w_H. The double heterozygote genotypes are of special interest since they can produce recombinant gametes.

	AB	**Ab**	**aB**	**ab**
AB	w_{11}	w_{12}	w_{13}	w_{14}
Ab	w_{21}	w_{22}	w_{23}	w_{24}
aB	w_{31}	w_{32}	w_{33}	w_{34}
ab	w_{41}	w_{42}	w_{43}	w_{44}

	BB	**Bb**	**bb**
AA	w_{11}	w_{12}	w_{22}
Aa	w_{13}	w_H	w_{24}
aa	w_{33}	w_{34}	w_{44}

Table 7.3 Expected frequencies of gametes under viability selection for two diallelic loci in a randomly mating population with a recombination rate of r between the loci. The expected gamete frequencies assume that the same gamete coming from either parent will have the same fitness in a progeny genotype (e.g. $w_{12} = w_{21}$). Eight genotypes have non-recombinant and recombinant gametes that are identical and so do not require a term for the recombination rate. Two genotypes produce novel recombinant gametes, requiring inclusion of the recombination rate to predict gamete frequencies. Summing down each column of the table gives the total frequency of each gamete in the next generation due to mating and recombination.

			Frequency of gametes in next generation			
Genotype	**Fitness**	**Total frequency**	**AB**	**Ab**	**aB**	**ab**
AB/AB	w_{11}	x_1^2	x_1^2			
AB/Ab	w_{12}	$2x_1x_2$	x_1x_2	x_1x_2		
AB/aB	w_{13}	$2x_1x_3$	x_1x_3		x_1x_3	
AB/ab	w_{14}	$2x_1x_4$	$(1-r)x_1x_4$	$(r)x_1x_4$	$(r)x_1x_4$	$(1-r)x_1x_4$
Ab/Ab	w_{22}	x_2^2		x_2^2		
Ab/aB	w_{23}	$2x_2x_3$	$(r)x_2x_3$	$(1-r)x_2x_3$	$(1-r)x_2x_3$	$(r)x_2x_3$
Ab/ab	w_{24}	$2x_2x_4$		x_2x_3	x_2x_3	
aB/aB	w_{33}	x_3^2			x_3^2	
aB/ab	w_{34}	$2x_3x_4$			x_3x_4	x_3x_4
ab/ab	w_{44}	x_4^2				x_4^2

Table 7.4 Fitness values based on the fecundities of mating pairs of male and female genotypes for a diallelic locus along with the expected genotype frequencies in the progeny of each possible male and female mating pair weighted by the fecundity of each mating pair. The frequencies of the AA, Aa, and aa genotypes are represented by *X*, *Y*, and *Z* respectively.

Male genotype	Female genotype . . .	Fitness value		
		AA	Aa	aa
AA		f_{11}	f_{12}	f_{13}
Aa		f_{21}	f_{23}	f_{23}
aa		f_{31}	f_{32}	f_{33}

Parental mating	Fecundity	Total frequency	Expected progeny genotype frequency		
			AA	Aa	aa
AA × AA	f_{11}	X^2	X^2	0	0
AA × Aa	f_{12}	XY	$^1/_2XY$	$^1/_2XY$	0
AA × aa	f_{13}	XZ	0	XZ	0
Aa × AA	f_{21}	YX	$^1/_2YX$	$^1/_2YX$	0
Aa × Aa	f_{22}	Y^2	$Y^2/4$	$(2Y^2)/4$	$Y^2/4$
Aa × aa	f_{23}	YZ	0	$^1/_2YZ$	$^1/_2YZ$
aa × AA	f_{31}	ZX	0	ZX	0
aa × Aa	f_{32}	ZY	0	$^1/_2ZY$	$^1/_2ZY$
aa × aa	f_{33}	Z^2	0	0	Z^2

Table 8.1 Nucleotide diversity (π) estimates reported from comparative studies of DNA sequence polymorphism from a variety of organisms and loci. All estimates are the average pairwise nucleotide differences per nucleotide site. For example, a value of $\pi = 0.02$ means that two in 100 sites vary between all pairs of DNA sequences in a sample.

Species	Locus	π	Reference
Drosophila melanogaster	*anon*1A3	0.0044	Andolfatto 2001
	Boss	0.0170	
	transformer	0.0051	
Drosophila simulans	*anon*1A3	0.0062	
	Boss	0.0510	
	transformer	0.0252	
Caenorhabditis elegans[a]	*tra*-2	0.0	Graustein et al. 2002
	glp-1	0.0009	
	COII	0.0102	
Caenorhabditis remanei[b]	*tra*-2	0.0112	
	glp-1	0.0188	
	COII	0.0228	
Arabidopsis thaliana[a]	*CAUL*	0.0042	Wright et al. 2003
	ETR1	0.0192	
	RbcL	0.0012	
Arabidopsis lyrata ssp. *Petraea*[b]	*CAUL*	0.0135	
	ETR1	0.0276	
	RbcL	0.0013	

[a]Mates by self-fertilization.
[b]Mates by outcrossing.

Table 8.3 Mean and variance in the number of substitutions at a neutral locus for the cases of divergence between two species and polymorphism within a single panmictic population. The rate of divergence is modeled as a Poisson process so the mean is identical to the variance. The mutation rate is μ and the $\theta = 4N_e\mu$. Refer to Fig. 8.17 for an illustration of divergence and ancestral polymorphism.

	Expected value or mean	Variance
Ancestral polymorphism	θ	$\theta + \theta^2$
Divergence	$2t\mu$	$2t\mu$
Sum	$2t\mu + \theta$	$2t\mu + \theta + \theta^2$

Table 8.4 Number of substitutions per nucleotide site observed over 49 nuclear genes for different orders of mammals. Divergences are divided into those observed at synonymous and nonsynonymous sites. Primates and artiodactyls (hoofed mammals such as cattle, deer, and pigs with an even number of digits) have longer generation times than do rodents. There were a total of 16,747 synonymous sites and 40,212 nonsynonymous sites. Data from Ohta (1995).

Mammalian group	Synonymous sites	Nonsynonymous sites
Primates	0.137	0.037
Artiodactyls	0.184	0.047
Rodents	0.355	0.062

Table 8.5 Estimates of polymorphism and divergence for two loci sampled from two species that form the basis of the HKA test. (a) The correlation of polymorphism and divergence under neutrality results in a constant ratio of divergence and polymorphism between loci independent of their mutation rate as well as a constant ratio of polymorphism or divergence between loci. (b) An illustration of ideal polymorphism and divergence estimates that would be consistent with the neutral null model. (c) Data for the *Adh* gene and flanking region (Hudson et al. 1987) is not consistent with the neutral model of sequence evolution because there is more *Adh* polymorphism within *Drosophila melanogaster* than expected relative to flanking region divergence between *D. melanogaster* and *D. sechellia*.

(a) Neutral case expectations

	Test locus	**Neutral reference locus**	**Ratio (test/reference)**
Focal species polymorphism (π)	$4N_e\mu_T$	$4N_e\mu_R$	$\frac{4N_e\mu_T}{4N_e\mu_R} = \frac{\mu_T}{\mu_R}$
Divergence between species (K)	$2T\mu_T$	$2T\mu_R$	$\frac{2T\mu_T}{2T\mu_R} = \frac{\mu_T}{\mu_R}$
Ratio (π/K)	$\frac{4N_e\mu_T}{2T\mu_T} = \frac{4N_e}{2T}$	$\frac{4N_e\mu_R}{2T\mu_R} = \frac{4N_e}{2T}$	

(b) Neutral case illustration

	Test locus	**Neutral reference locus**	**Ratio (test/reference)**
Focal species polymorphism (π)	0.10	0.25	0.40
Divergence between species (K)	0.05	0.125	0.40
Ratio (π/K)	2.0	2.0	

(c) Empirical data from *D. melanogaster* and *D. sechellia*

	Adh	***5′ Adh* flanking region**	**Ratio (Adh/flank)**
D. melanogaster polymorphism (π)	0.101	0.022	4.59
Between species divergence (K)	0.056	0.052	1.08
Ratio (π/K)	1.80	0.42	

Table 8.6 Estimates of polymorphism and divergence (fixed sites) for nonsynonymous and synonymous sites at a coding locus form the basis of the MK test. (a) Under neutrality, the number of nonsynonymous sites divided by the number of synonymous sites is equal to the ratio of the nonsynonymous and synonymous mutation rates. This ratio should be constant both for nucleotide sites with fixed differences between species and polymorphic sites within the species of interest. (b) An illustration of ideal nonsynonymous and synonymous site changes that would be consistent with the neutral null model. (c) Data for the *Adh* locus in *D. melanogaster* (McDonald & Kreitman 1991) show an excess of *Adh* nonsynonymous polymorphism compared with that expected based on divergence. (d) Data for the *Hla*-B locus for humans show an excess of polymorphism and more nonsynonymous than synonymous changes, consistent with balancing selection (Garrigan & Hedrick 2003).

	Fixed differences	**Polymorphic sites**
(a) Neutral case expectations		
Nonsynonymous sites (N)	$N_F = 2T\mu_N$	$N_P = 4N_e\mu_N$
Synonymous sites (S)	$S_F = 2T\mu_S$	$S_P = 4N_e\mu_S$
Ratio (N/S)	$\frac{N_F}{S_F} = \frac{2T\mu_N}{2T\mu_S} = \frac{\mu_N}{\mu_S}$	$\frac{N_P}{S_P} = \frac{4N_e\mu_N}{4N_e\mu_S} = \frac{\mu_N}{\mu_S}$
(b) Neutral case illustration		
Nonsynonymous changes	4	15
Synonymous changes	12	45
Ratio	0.33	0.33
(c) Empirical data from *Adh* locus for *D. melanogaster* (McDonald & Kreitman 1991)		
Nonsynonymous changes	2	7
Synonymous changes	42	17
Ratio	0.045	0.412
(d) Empirical data for the *Hla*-B locus for humans (Garrigan & Hedrick 2003)		
Nonsynonymous changes	0	76
Synonymous changes	0	49
Ratio	–	1.61

Table 9.1 Symbols commonly used to refer to categories or causes of variation in quantitative traits. Variation is indicated by *V* while the specific cause of that variation is indicated by a subscript capital letter (with one exception). Total genetic variation (V_G) in phenotype can be divided into three subcategories.

Symbol	Definition
V_P	Total variance in a quantitative trait or phenotype
V_G	Variance in phenotype due to all genetic causes
V_A	Variance in phenotype caused by additive genetic variance or the effects of alleles
V_D	Variance in phenotype caused by dominance genetic variance or deviations from additive values due to dominance
V_I	Variance in phenotype caused by interaction genetic variance (epistasis between and among loci)
V_E	Variance in phenotype caused by environmental variation
$V_{G\times E}$	Variance in phenotype caused by genotype-by-environment interaction
V_{Ec}	Variance in phenotype caused by environmental variation shared in common by parents and offspring or by relatives

Table 9.2 The eight uncorrelated (or orthogonal) types of genetic effects that can occur between two diallelic loci. Four of the eight types of genetic effects are interactions that give rise to V_I. The genotypic values assume all allele frequencies are $^1/_2$. Table after Goodnight (2000).

Genetic effect	Genotypes and phenotypes . . .	Genotypic value		
		A_1A_1	A_1A_2	A_2A_2
Additive A locus	B_1B_1	1	0	–1
	B_1B_2	1	0	–1
	B_2B_2	1	0	–1
Additive B locus	B_1B_1	1	1	1
	B_1B_2	0	0	0
	B_2B_2	–1	–1	–1
A locus dominance	B_1B_1	0	1	0
	B_1B_2	0	1	0
	B_2B_2	0	1	0
B locus dominance	B_1B_1	0	0	0
	B_1B_2	1	1	1
	B_2B_2	0	0	0
Additive-by-additive interaction	B_1B_1	1	0	–1
	B_1B_2	0	0	0
	B_2B_2	–1	0	1
Additive (A locus)-by-dominance (B locus) interaction	B_1B_1	1	–1	1
	B_1B_2	0	0	0
	B_2B_2	–1	1	–1
Dominance (A locus)-by-additive (B locus) interaction	B_1B_1	1	0	–1
	B_1B_2	–1	0	1
	B_2B_2	1	0	–1
Dominance-by-dominance interaction	B_1B_1	–1	1	–1
	B_1B_2	1	–1	1
	B_2B_2	–1	1	–1

Additive gene action

Genotypes	BB	Bb	bb
Phenotypes	3	2	1

	Cross	Mean phenotype
(a)		
Parents	BB × bb	$\frac{3+1}{2} = 2$
Progeny	Bb	2
(b)		
Parents	Bb × Bb	2
Progeny	$^1/_4$ BB, $^1/_2$ Bb, $^1/_4$ bb	$^1/_4(3) + {}^1/_2(2) + {}^1/_4(1) = 2$

Complete dominance

Genotypes	BB	Bb	bb
Phenotypes	3	3	1

	Cross	Mean phenotype
(a)		
Parents	BB × bb	$\frac{3+1}{2} = 2$
Progeny	Bb	3
(b)		
Parents	Bb × Bb	3
Progeny	$^1/_4$ BB, $^1/_2$ Bb, $^1/_4$ bb	$^1/_4(3) + {}^1/_2(3) + {}^1/_4(1) = 2.5$

Table 9.3 Examples of parental and progeny mean phenotypes that illustrate the impacts of additive gene action (top) or complete dominance gene action (bottom). For both types of gene action, the phenotypic value of each genotype is given and the genotypes of two possible parental crosses are shown along with the genotypes in the progeny from each cross. Under additive gene action the mean phenotypic values are identical in the parents and progeny because phenotypic values are a function of allele frequencies and alleles are identical in parents and progeny. In contrast, under complete dominance parent and progeny mean phenotypic values differ because phenotypic values are a function of the genotype and genotype frequencies differ between parents and progeny.

Table 9.4 Examples of response to selection for two phenotypes with the possibility of phenotypic or additive genetic covariance. The elements of the phenotypic variance/covariance matrix (***P***), the additive genetic variance/covariance matrix (***G***), the vector of selection differentials (***s***), and the vector of predicted changes in mean phenotype ($\Delta\bar{\mathbf{z}}$) are shown in (a).

(a)

G

	[Trait A]	**[Trait B]**
Trait A	h^2	Genetic cov(A, B)
Trait B	Genetic cov(A, B)	h^2

P

	[Trait A]	**[Trait B]**
Trait A	Variance(A)	Phenotypic cov(A, B)
Trait B	Phenotypic cov(A, B)	Variance(B)

s = [selection differential trait A, selection differential trait B]
$\Delta\bar{\mathbf{z}}$ = [change in mean of trait A, change in mean of trait B]

(b)

$\boldsymbol{G} = \begin{matrix} 0.5 & 0 \\ 0 & 0.5 \end{matrix}$ $\boldsymbol{P} = \begin{matrix} 1.0 & 0 \\ 0 & 1.5 \end{matrix}$

$\boldsymbol{s} = 0.5, 0.5$

$\Delta\bar{\mathbf{z}} = 0.25, 0.1667$

(c)

$\boldsymbol{G} = \begin{matrix} 0.5 & 0 \\ 0 & 0.5 \end{matrix}$ $\boldsymbol{P} = \begin{matrix} 1.0 & 0.6 \\ 0.6 & 1.5 \end{matrix}$

$\boldsymbol{s} = 0.5, 0$

$\Delta\bar{\mathbf{z}} = 0.3289, -0.1316$

(d)

$\boldsymbol{G} = \begin{matrix} 0.5 & 0.6 \\ 0.6 & 0.5 \end{matrix}$ $\boldsymbol{P} = \begin{matrix} 1.0 & 0 \\ 0 & 1.5 \end{matrix}$

$\boldsymbol{s} = 0.5, 0$

$\Delta\bar{\mathbf{z}} = 0.25, 0.20$

Table 9.5 Derivation of the expected phenotypic value for the three marker-locus genotypes when QTL mapping with a single marker locus associated with a single QTL. The three genetic marker genotypes may be associated with any of the three possible QTL genotypes because of recombination during the formation of F2 gametes. The difference between the M_1/M_1 and M_2/M_2 marker class means (expressions in the Marker-class mean value column) is equal to $2\hat{a}$. The phenotypic value of each marker locus genotype is a function of both the additive and dominance effects of the QTL (a and d) as well as the recombination rate (r). So unless there is no dominance and no recombination, estimates of QTL effects from single-marker-locus mapping are always minimum estimates. The gametes and expected gamete frequencies are given in Fig. 9.15.

Gametes	F2 genotype	Genotype frequency	Genotypic value	Frequency-weighted genotypic value	Marker genotype	Marker-class contribution to F2 population mean value	Marker genotype frequency in F2 population	Marker-class mean value
c/c	$\frac{M_1Q_1}{M_1Q_1}$	$\left(\frac{1-r}{2}\right)^2$	$+a$	$a\left(\frac{1-r}{2}\right)^2$	$\frac{M_1}{M_1}$	$\bar{G}^{pop}_{M_1M_1} = \frac{a(1-2r)}{4} + \frac{2dr(1-r)}{4}$	$^1/_4$	$\bar{G}_{M_1M_1} = a(1-2r) + 2dr(1-r)$
c/r	$\frac{M_1Q_1}{M_1Q_2}$	$(2)\frac{r}{2}\left(\frac{1-r}{2}\right)$	d	$2d\frac{r}{2}\left(\frac{1-r}{2}\right)$				
r/r	$\frac{M_1Q_2}{M_1Q_2}$	$\left(\frac{r}{2}\right)^2$	$-a$	$-a\left(\frac{r}{2}\right)^2$				
c/c	$\frac{M_1Q_1}{M_2Q_2}$	$(2)\left(\frac{1-r}{2}\right)^2$	d	$2d\left(\frac{1-r}{2}\right)^2$	$\frac{M_1}{M_2}$	$\bar{G}^{pop}_{M_1M_1} = \frac{d[(1-r)^2 + r^2]}{2}$	$^1/_2$	$\bar{G}_{M_1M_2} = d[(1-r)^2 + r^2]$
r/r	$\frac{M_1Q_2}{M_2Q_1}$	$(2)\left(\frac{r}{2}\right)^2$	d	$2d\left(\frac{r}{2}\right)^2$				
c/r	$\frac{M_1Q_1}{M_2Q_1}$	$(2)\frac{r}{2}\left(\frac{1-r}{2}\right)$	$+a$	$2a\frac{r}{2}\left(\frac{1-r}{2}\right)$				
r/c	$\frac{M_1Q_2}{M_2Q_2}$	$(2)\frac{r}{2}\left(\frac{1-r}{2}\right)$	$-a$	$-2a\frac{r}{2}\left(\frac{1-r}{2}\right)$				
c/c	$\frac{M_2Q_2}{M_2Q_2}$	$\left(\frac{1-r}{2}\right)^2$	$-a$	$-a\left(\frac{1-r}{2}\right)^2$	$\frac{M_2}{M_2}$	$\bar{G}^{pop}_{M_2M_2} = \frac{-a(1-2r)}{4} + \frac{2dr(1-r)}{4}$	$^1/_4$	$\bar{G}_{M_2M_2} = -a(1-2r) + 2dr(1-r)$
c/r	$\frac{M_2Q_2}{M_2Q_1}$	$(2)\frac{r}{2}\left(\frac{1-r}{2}\right)$	d	$2d\frac{r}{2}\left(\frac{1-r}{2}\right)$				
r/r	$\frac{M_2Q_1}{M_2Q_1}$	$\left(\frac{r}{2}\right)^2$	$+a$	$a\left(\frac{r}{2}\right)^2$				

Table 9.6 Derivation of the expected phenotypic value for two genetic marker genotypes when QTL mapping with pairs of marker loci that flank a QTL. There are a total of nine genetic marker genotypes possible with two genetic marker loci. Like QTL mapping based on a single genetic marker, the phenotypic value of the $A_1A_1B_1B_1$ marker genotype is a function of both the additive and dominance effects of the QTL. In contrast, the expected phenotypic value of the $A_1A_2B_1B_2$ marker genotype is a function only of *d*. Therefore, the $A_1A_2B_1B_2$ marker class mean value provides an estimate of the dominance coefficient independent of *a*. The gametes and expected gamete frequencies are given in Fig. 9.16.

Marker genotype	Marker genotype frequency	F2 genotype	F2 genotype frequency	F2 genotypic value	Frequency-weighted F2 genotypic value
$A_1A_1B_1B_1$	$\frac{(1-r)^2}{4}$	$\frac{A_1Q_1B_1}{A_1Q_1B_1}$	$\left(\frac{(1-r_A)(1-r_B)}{2}\right)^2$	$+a$	$\frac{a(1-r_A)^2(1-r_B)^2}{4}$
		$\frac{A_1Q_1B_1}{A_1Q_2B_1}$	$2\left(\frac{(1-r_A)(1-r_B)}{2}\right)\left(\frac{r_Ar_B}{2}\right)$	d	$\frac{2dr_Ar_B(1-r_A)(1-r_B)}{4}$
		$\frac{A_1Q_2B_1}{A_1Q_2B_1}$	$\left(\frac{r_Ar_B}{2}\right)^2$	$-a$	$\frac{-ar_A^2r_B^2}{4}$
$\bar{G}^{pop}_{A_1A_1B_1B_1} = \frac{a(1-r_A)^2(1-r_B)^2}{4} + \frac{2dr_Ar_B(1-r_A)(1-r_B)}{4} + \frac{-ar_A^2r_B^2}{4}$					
$\bar{G}_{A_1A_1B_1B_1} = \frac{4}{(1-r)^2}\left[\frac{a(1-r_A)^2(1-r_B)^2}{4} + \frac{2dr_Ar_B(1-r_A)(1-r_B)}{4} + \frac{-ar_A^2r_B^2}{4}\right] = a\frac{(1-r_A)^2(1-r_B)^2 - r_A^2r_B^2}{(1-r)^2} + d\frac{2r_Ar_B(1-r_A)(1-r_B)}{(1-r)^2}$					
$A_1A_2B_1B_2$	$\frac{(1-r)^2+r^2}{4}$	$\frac{A_1Q_1B_1}{A_2Q_2B_2}$	$\left(\frac{(1-r_A)(1-r_B)}{2}\right)^2$	d	$\frac{d(1-r_A)^2(1-r_B)^2}{4}$
		$\frac{A_1Q_1B_2}{A_2Q_2B_1}$	$\left(\frac{(1-r_A)r_B}{2}\right)^2$	d	$\frac{d(1-r_A)^2r_B^2}{4}$
		$\frac{A_2Q_1B_1}{A_1Q_1B_2}$	$\left(\frac{r_A(1-r_B)}{2}\right)^2$	d	$\frac{dr_A^2(1-r_B)^2}{4}$
		$\frac{A_1Q_2B_1}{A_2Q_1B_2}$	$\left(\frac{r_Ar_B}{2}\right)^2$	d	$\frac{dr_A^2r_B^2}{4}$
		$\frac{A_1Q_1B_1}{A_2Q_1B_2}$	$2\left(\frac{(1-r_A)(1-r_B)}{2}\right)\left(\frac{r_Ar_B}{2}\right)$	$+a$	$\frac{2ar_Ar_B(1-r_A)(1-r_B)}{4}$
		$\frac{A_2Q_2B_2}{A_1Q_2B_1}$	$2\left(\frac{(1-r_A)(1-r_B)}{2}\right)\left(\frac{r_Ar_B}{2}\right)$	$-a$	$\frac{-2ar_Ar_B(1-r_A)(1-r_B)}{4}$
$\bar{G}^{pop}_{A_1A_2B_1B_2} = \frac{d(1-r_A)^2(1-r_B)^2}{4} + \frac{d(1-r_A)^2r_B^2}{4} + \frac{dr_A^2(1-r_B)^2}{4} + \frac{dr_A^2r_B^2}{4} + \frac{2ar_Ar_B(1-r_A)(1-r_B)}{4} + \frac{-2ar_Ar_B(1-r_A)(1-r_B)}{4}$					
$\bar{G}_{A_1A_2B_1B_2} = \frac{4}{(1-r)^2+r^2}\left[\frac{d(1-r_A)^2(1-r_B)^2}{4} + \frac{d(1-r_A)^2r_B^2}{4} + \frac{dr_A^2(1-r_B)^2}{4} + \frac{dr_A^2r_B^2}{4}\right] = d\frac{r_Ar_B + r_A(1-r_B)^2 + (1-r_A)r_B^2 + (1-r_A)(1-r_B)}{r^2+(1-r)^2}$					

Table 9.7 Examples of QTLs identified by mapping with genetic marker loci.

Organism	Phenotype	Number of marker loci	Number of QTLs	Reference
Arabidopsis thaliana	Days to first flower	65	7	Kearsey et al. 2003
	Number of buds at flowering		28	
	Rosette size at 21 days		4	
	Rosette size at flowering		10	
Dogs	Body size	116	1	Sutter et al. 2007
Drosophila santomea × *D. yakuba*	Prezygotic reproductive isolation	32	6	Moehring et al. 2006
Humans	Taste sensitivity to PTC	50	1	Kim et al. 2003
	Stature	> 253	3	Perola et al. 2007
Louisiana irises	Flowering time	> 414	17	Martin et al. 2007
Stickleback fish	Bony plates	160	4	Colosimo et al. 2004
Zea mays	Kernel oil concentration	488	> 50	Laurie et al. 2004

PTC, phenylthiocarbamide.

Table 9.8 Derivation of the expected phenotypic values for marker genotypes used to estimate $\hat{a}$ in a P1 × F1 backcross mating design when QTL mapping with pairs of marker loci that flank a QTL.

F1 parent gamete	F1 gamete frequency	BC progeny genotype	BC progeny genotypic value	Frequency-weighted genotypic value	BC progeny marker genotype	Marker genotype frequency	Marker class mean ($\bar{G}^{BC}_{A_xA_xB_xB_x}$)
$A_1Q_1B_1$	$\frac{(1-r)}{2}$	$\frac{A_1Q_1B_1}{A_1Q_1B_1}$	$+a$	$a\frac{(1-r)}{2}$	$A_1A_1B_1B_1$	$\frac{(1-r)}{2}$	a
$A_1Q_1B_2$	$\frac{(1-r_A)r_B}{2}$	$\frac{A_1Q_1B_1}{A_1Q_1B_2}$	$+a$	$\frac{ar_B + ar_Ar_B + dr_A - dr_Ar_B}{2} \approx \frac{ar_B + dr_A}{2}$	$A_1A_1B_1B_2$	$\frac{r}{2}$	$\frac{ar_B + dr_A}{r}$
$A_1Q_2B_2$	$\frac{r_A(1-r_B)}{2}$	$\frac{A_1Q_1B_1}{A_1Q_2B_2}$	d				
$A_2Q_2B_1$	$\frac{(1-r_A)r_B}{2}$	$\frac{A_1Q_1B_1}{A_2Q_2B_1}$	d	$\frac{ar_A + ar_Ar_B + dr_B - dr_Ar_B}{2} \approx \frac{ar_A + dr_B}{2}$	$A_1A_2B_1B_1$	$\frac{r}{2}$	$\frac{ar_A + dr_B}{r}$
$A_2Q_1B_1$	$\frac{r_A(1-r_B)}{2}$	$\frac{A_1Q_1B_1}{A_2Q_1B_1}$	$+a$				
$A_2Q_2B_2$	$\frac{(1-r)}{2}$	$\frac{A_1Q_1B_1}{A_2Q_2B_2}$	d	$d\frac{(1-r)}{2}$	$A_1A_2B_1B_2$	$\frac{(1-r)}{2}$	d

Table 10.1 The population mean phenotype (M) obtained from genotype frequencies under random mating, genotypic values, and frequency-weighted genotypic values for a diallelic locus. These expectations assume that the environmental deviation is zero for each genotype.

Genotype	Frequency	Genotypic value	Frequency-weighted genotypic value
A_1A_1	p^2	a	p^2a
A_1A_2	$2pq$	d	$2pqd$
A_2A_2	q^2	$-a$	$-q^2a$
			$M = a(p - q) + 2pqd$

Table 10.2 The mean value of all genotypes that contain either an A_1 (M_{A_1}) or an A_2 (M_{A_2}) allele. The average effect of an allele (α_x) is the difference between the mean value of the genotypes that contain a given allele and the population mean ($\alpha_x = M_{A_x} - M$).

Allele	Genotype value			Mean value of all genotypes containing a given allele
	A_1A_1	A_1A_2	A_2A_2	
	$+a$	d	$-a$	
A_1	p	q	0	$M_{A_1} = pa + qd$
A_2	0	p	q	$M_{A_2} = pd - qa$

(a) $d = 0.0$, $p = 0.5$, $q = 0.5$
$M = 10.5(0.5 - 0.5) + 2(0.5)(0.5)(0.0) = 0.0$
A_1 $M_{A_1} = pa + qd = (0.5)(10.5) + (0.5)(0.0) = 5.25$
$\alpha_1 = M_{A_1} - M = 5.25 - 0.0 = 5.25$
$\alpha_1 = q(a + d(q - p)) = 0.5(10.5 + 0.0(0.5 - 0.5)) = 5.25$
$\alpha = a + d(q - p) = 10.5 + 0.0(0.5 - 0.5) = 10.5$
$\alpha_1 = q\alpha = (0.5)(10.5) = 5.25$

(b) $d = 0.0$, $p = 0.9$, $q = 0.1$
$M = 10.5(0.9 - 0.1) + 2(0.9)(0.1)(0.0) = 8.4$
A_1 $M_{A_1} = pa + qd = (0.9)(10.5) + (0.1)(0.0) = 9.45$
$\alpha_1 = M_{A_1} - M = 9.45 - 8.4 = 1.05$
$\alpha_1 = q(a + d(q - p)) = 0.1(10.5 + 0.0(0.1 - 0.9)) = 1.05$
$\alpha = a + d(q - p) = 10.5 + 0.0(0.1 - 0.9) = 10.5$
$\alpha_1 = q\alpha = (0.1)(10.5) = 1.05$

(c) $d = 5.25$, $p = 0.5$, $q = 0.5$
$M = 10.5(0.5 - 0.5) + 2(0.5)(0.5)(5.25) = 2.625$
A_1 $M_{A_1} = pa + qd = (0.5)(10.5) + (0.5)(5.25) = 7.875$
$\alpha_1 = M_{A_1} - M = 7.875 - 2.625 = 5.25$
$\alpha_1 = q(a + d(q - p)) = 0.5(10.5 + 5.25(0.5 - 0.5)) = 5.25$
$\alpha = a + d(q - p) = 10.5 + 5.25(0.5 - 0.5) = 10.5$
$\alpha_1 = q\alpha = (0.5)(10.5) = 5.25$

(d) $d = 5.25$, $p = 0.9$, $q = 0.1$
$M = 10.5(0.9 - 0.1) + 2(0.9)(0.1)(5.25) = 9.345$
A_1 $M_{A_1} = pa + qd = (0.9)(10.5) + (0.1)(5.25) = 9.975$
$\alpha_1 = M_{A_1} - M = 9.975 - 9.345 = 0.630$
$\alpha_1 = q(a + d(q - p)) = 0.1(10.5 + 5.25(0.1 - 0.9)) = 0.630$
$\alpha = a + d(q - p) = 10.5 + 5.25(0.1 - 0.9) = 6.3$
$\alpha_1 = q\alpha = (0.1)(6.3) = 0.63$

Table 10.3 Examples of the average effect for the *IGF*1 locus in dogs. All cases assume that $a = 10.5$ kg as shown in the genotypic scale in Fig. 10.1. For each set of allele frequencies and dominance, the table shows the population mean (M), the mean value of all genotypes that contain an A_1 allele (M_{A_2}), the average effect of an allelic replacement (α), and the average effect of an A_1 allele (α_1). Values are all in kilograms and relative to the midpoint value of 19.5 kg.

Table 10.4 The mean phenotypic value of progeny that result when an individual of the genotype A_1A_1 mates randomly. All genotypes in the population have Hardy–Weinberg expected frequencies. Therefore, each of the mating pairs has an expected frequency of p^2, $2pq$, or q^2. The mean value of all progeny produced by the A_1A_1 genotype is the frequency-weighted sum of the progeny phenotypic values. $M_{\text{progeny } A_1A_1}$ forms the basis of the breeding value since the breeding value for A_1A_1 is $M_{\text{progeny } A_1A_1} - M$.

Focal genotype	A_1A_1			
Mate genotypes	A_1A_1		A_1A_2	A_2A_2
Mating frequency	p^2		$2pq$	q^2
Progeny genotype and relative frequency from each mating	A_1A_1	$^1/_2$ A_1A_1	$^1/_2$ A_1A_2	A_1A_2
Progeny values	$+a$	$+a$	d	d
Progeny mean value	$M_{\text{progeny } A_1A_1} = p^2a + 2pq(^1/_2a + {}^1/_2d) + q^2d = ap + dq$			

Table 10.5 Examples of breeding values for the three *IGF*1 locus genotypes in dogs. Values are all in kilograms and relative to the midpoint value of 19.5 kg.

	Breeding value		
	A_1A_1	A_1A_2	A_2A_2
(a) $d = 0.0$, $p = 0.5$, $q = 0.5$, $M = 0.0$, $\alpha = 10.5$	2(0.5)(10.5) = 10.5	(0.5 – 0.5)(10.5) = 0.0	–2(0.5)(10.5) = –10.5
(b) $d = 0.0$, $p = 0.9$, $q = 0.1$, $M = 8.4$, $\alpha = 10.5$	2(0.1)(10.5) = 2.1	(0.1 – 0.9)(10.5) = –8.4	–2(0.9)(10.5) = –18.9
(c) $d = 5.25$, $p = 0.5$, $q = 0.5$, $M = 2.625$, $\alpha = 10.5$	2(0.5)(10.5) = 10.5	(0.5 – 0.5)(10.5) = 0.0	–2(0.5)(10.5) = –10.5
(d) $d = 5.25$, $p = 0.9$, $q = 0.1$, $M = 9.345$, $\alpha = 6.3$	2(0.1)(6.3) = 1.26	(0.1 – 0.9)(6.3) = –5.04	–2(0.9)(6.3) = –11.34

Table 10.6 Expressions for genotypic values relative to the population mean, breeding values and dominance deviations. Genotypic values can be expressed relative to the population mean by subtracting the population mean ($M = a(p - q) + 2pqd$) from a genotypic value measured relative to the midpoint. The dominance deviation is the difference between the genotypic value expressed relative to the population mean (M) and the breeding value.

	Value		
Genotype . . .	**A_1A_1**	**A_1A_2**	**A_2A_2**
Genotypic value relative to midpoint	$+a$	d	$-a$
Genotypic value relative to population mean	$2q(a - dp)$ $2q(\alpha - qd)$	$a(p + q) + d(1 - 2pq)$ $(q - p)\alpha + 2pqd$	$-2p(a - dp)$ $-2p(\alpha + pd)$
Breeding value	$2q(a + d(q - p))$ $2q\alpha$	$(q - p)(a + d(q - p))$ $(q - p)\alpha$	$-2p(a + d(q - p))$ $-2p\alpha$
Dominance deviation	$-2q^2d$	$2pqd$	$-2p^2d$

Table 10.7 Genotypic values, breeding values, and dominance deviations for the three *IGF*1 locus genotypes in dogs. Genotypic values, breeding value and dominance deviation values are all given relative to the population mean, *M*. All values are in kilograms.

	Genotype		
	A_1A_1	A_1A_2	A_2A_2
(a) $d = 0.0$, $p = 0.5$, $q = 0.5$, $M = 0.0$, $\alpha = 10.5$			
Genotype frequency	0.25	0.5	0.25
Genotypic value	10.5	0.0	–10.5
Breeding value	10.5	0.0	–10.5
Dominance deviation	0.0	0.0	0.0
(b) $d = 0.0$, $p = 0.9$, $q = 0.1$, $M = 8.4$, $\alpha = 10.5$			
Genotype frequency	0.81	0.18	0.01
Genotypic value	2.1	–8.4	–10.5
Breeding value	2.1	–8.4	–10.5
Dominance deviation	0.0	0.0	0.0
(c) $d = 5.25$, $p = 0.5$, $q = 0.5$, $M = 2.625$, $\alpha = 10.5$			
Genotype frequency	0.25	0.5	0.25
Genotypic value	7.875	2.625	–13.125
Breeding value	10.5	0.0	–10.5
Dominance deviation	–2.625	–2.625	–2.625
(d) $d = 5.25$, $p = 0.9$, $q = 0.1$, $M = 9.345$, $\alpha = 6.3$			
Genotype frequency	0.81	0.18	0.01
Genotypic value	1.155	–4.095	–19.845
Breeding value	1.26	–5.04	–11.34
Dominance deviation	–0.105	0.945	–8.505

Table 10.8 Expected covariance in genotypic values between groups of relatives.

Relatives		Covariance in genotypic values
Offspring (x)	One parent (y)	$^1/_2V_A$
Offspring (x)	Mid-parent (y)	$^1/_2V_A$
Half siblings		$^1/_4V_A$
Full siblings		$^1/_2V_A + {}^1/_4V_D$
Nephew/niece (x)	Uncle/aunt (y)	$^1/_4V_A$
First cousins		$^1/_8V_A$
Monozygotic twins		$V_A + V_D$

Table 10.9 Frequencies and mean values for parents and progeny used to derive the covariance between the average value of parents (mid-parent value) and the average value of the progeny from each parental mating.

Parental mating	Parental mating frequency	Mid-parent value ($\bar{P}_i$)	Progeny genotype frequencies			Progeny value (O_i)
			A_1A_1	A_1A_2	A_2A_2	
$A_1A_1 \times A_1A_1$	p^4	a	1	–	–	a
$A_1A_1 \times A_1A_2$	$4p^3q$	$\frac{1}{2}(a + d)$	$\frac{1}{2}$	$\frac{1}{2}$	–	$\frac{1}{2}(a + d)$
$A_1A_1 \times A_2A_2$	$2p^2q^2$	$a + (-a) = 0$	–	1	–	$a + (-a) = 0$
$A_1A_2 \times A_1A_2$	$4p^2q^2$	d	$\frac{1}{4}$	$\frac{1}{2}$	$\frac{1}{4}$	$\frac{1}{2}d$
$A_1A_2 \times A_2A_2$	$4pq^3$	$\frac{1}{2}(-a + d)$	–	$\frac{1}{2}$	$\frac{1}{2}$	$\frac{1}{2}(-a + d)$
$A_2A_2 \times A_2A_2$	q^4	$-a$	–	–	1	$-a$